Blessing Way Midwifery

Prenatal Testing Options

Women face many choices in their pregnancy and want to make decisions that are best for themselves, their baby and their family. The decision to proceed with any prenatal testing is a personal one. At your first appointment with the midwives a family history taken and a discussion will take place regarding the options available to you. This information is intended to provide you with some preliminary information about testing to assist you in gathering information and making a choice that is best for you.

Why do you take my family history?

Some families may have concerns about their family histories and risks to their pregnancy. Genetic counselling if available for women who have family history concerns and would like to understand their risks to their offspring.

There are two type of prenatal testing available. They include Prenatal *Screening* and Prenatal *Diagnostic testing*. An understanding of the differences between prenatal screening and prenatal diagnostic tests is important.

A *screen* is a test that provides you with *risk information* only.

It does not give you a 'yes' or 'no' answer. Rather, it provides information, specific to your pregnancy, about your chance of having a baby with a particular chromosome condition or birth defect. If your screen result places you at an 'increased' risk you will offered a diagnostic test. Prenatal Screening options are explained further below.

Prenatal *diagnostic tests* answers the question: "Does my baby have Down syndrome

Prenatal diagnostic tests include amniocentesis or chorionic villus sampling (CVS). These two tests provide you with a *diagnosis*, that is, a 'yes' or 'no' answer about chromosome conditions.

Prenatal Screening Options:

First Trimester:

Nuchal translucency screening:

The nuchal translucency screening uses prenatal ultrasound to measure the amount of fluid at the back of the baby's neck. This measurement, together with your age, is used to provide you with a personal risk of having a baby with Down syndrome or two other chromosome conditions called Trisomy 13 and Trisomy 18. The ultrasound is also used to ensure fetal viability (baby is alive), diagnosis twins (or greater!) ensure your due date is correct and detect some birth defects that are identifiable at this stage in the pregnancy.

Approximately 80% of pregnancies carrying a baby with Down syndrome are detected with nuchal translucency screening. Women who receive an 'increased risk result' (a risk greater than 1:300) are offered prenatal diagnosis. An increased nuchal translucency *may* also be present in babies at increased risk for a heart defect and a special heart ultrasound is offered at 20 weeks in those circumstances.

For the most part, women receive reassuring risk information, providing them with peace of mind early in their pregnancy. For women who are over 35 years of age and undecided about prenatal diagnosis, the risk evaluation may further assist them in making decisions about whether or not to proceed with amniocentesis.

Nuchal translucency Screening is currently available at the Southern Alberta Centre for Maternal Fetal Medicine by certified ultrasound technicians, who are directly supervised by Perinatal specialists. The parents are immediately informed of the results as well as their options. If the results are unfavorable, a genetic counselor may be available to provide further counseling.

First Trimester Combined Screening:

First Trimester Combined Screening involves a maternal blood test and nuchal translucency ultrasound between 11 weeks and 13 weeks 6 days in pregnancy. First Trimester Combined Screening has replaced nuchal translucency screening at the Southern Alberta Centre for Maternal Fetal Medicine as the detection rate is approximately 90%. For more information please see the educational video at www.earlyriskassessment.com

Second trimester:

Maternal Serum Screening:

Maternal serum Screening is a second trimester screen for Down syndrome, trisomy 18 and neural tube defects (spina bifida). The test involves a blood sample at 15-20 weeks in pregnancy. Three proteins are measured in the mother's blood that are combined with a woman's age to provide a risk estimate for having a baby with Down syndrome. The detection rate is 60-70%.

At present, it is encouraged to have only one Down syndrome screen. Therefore if you are considering first trimester screening, you would be asked not to have second trimester Maternal Serum Screening.

18-20 week ultrasound

The 18-20 week ultrasound serves to evaluate the growth and development of the baby. This ultrasound is used to evaluate the baby for serious birth defects and screen for physical features that may place a pregnancy at increased risk for chromosome conditions. Most women receive very reassuring information from their ultrasound. However, in a small proportion of women, a birth defect may be identified. In these cases, women will be offered counselling and the option of prenatal diagnosis.

There may be ultrasound findings that are of unknown significance that may cause parents much concern. These "soft markers" (such as echogenic foci and choroids plexus cysts) are *most often* seen as a variation in normally developing fetus' but may also be seen in babies with chromosomal abnormalities. If one of these soft markers is identified you will need to make a decision about whether further testing such as amniocentesis is right for you.

Prenatal Screening – What is important to consider:

With prenatal screening a cut-off is used to categorize risks as either 'positive' (increased) or 'negative' (reduced) risk results.

As with any screening, there will be women who receive 'positive' results and have a healthy normal baby. With First trimester Combined Screening, approximately 6 out of 100 women screening will receive a 'positive' result. With Maternal Serum Screening, about 7-9 out of 100 women will receive a 'positive' result. Of these women, the vast majority will have babies with normal chromosomes. In these cases, this would be called a false positive result.

There is always a small chance that a woman who receives a 'negative' results may have a baby with a chromosome condition like Down syndrome. This is called a false negative.

All screening tests have false positive and false negative rates. Many families find this 'false positive' rate unacceptable or anxiety-provoking which is something to consider before proceeding with testing.

Prenatal Diagnosis options

Amniocentesis:

Amniocentesis is medical procedure performed by a doctor that diagnoses *chromosome conditions* and identifies pregnancies at risk for neural tube defects (spina bifida).

Amniocentesis is generally performed between 15 and 18 weeks of pregnancy, although earlier or later amniocentesis is offered in some circumstances. This test is routinely offered to all women 35 years or older or women who have received

a positive screen test result.

The procedure is closely followed by ultrasound which maximizes safety for both the woman and her baby. Ultrasound is used to determine the position of the baby. A fine needle is carefully inserted through the woman's abdomen and a small amount (one to two tablespoons) of fluid is removed for testing.

The majority of amniocenteses are performed without complications. There is, however, a risk of miscarriage associated with the test. For every 200 amniocenteses performed, one pregnancy (0.5%) will be lost to miscarriage due to the procedure.

Chorionic Villus sampling:

Chorionic Villus Sampling (CVS) is a diagnostic test for chromosome conditions performed between 11 and 13 weeks in pregnancy. Under the guidance of ultrasound a thin instrument is inserted into the vagina and through the cervix to reach the chorionic villi (the early placenta). A small sample of chorionic villi is removed from the developing placenta. Chromosome testing is performed on this sample.

The risk of miscarriage associated with this test is approximately 1%. In Calgary, CVS is currently reserved for women with specific prenatal indications.

18-20 week Detailed OB Scan:

The second trimester ultrasound is both a screening and diagnostic test. It screens for physical signs in the baby suggestive of a chromosome condition, but is diagnostic for a number of birth defects such as congenital heart defects and spina bifida.

Should a birth defect or chromosome condition be detected in pregnancy

Genetic counselling is available to review the results, answer your questions and ensure you have sufficient information to help you to make decisions about whether to continue your pregnancy or to feel better prepared to parent a child with special needs.

Take home messages

• Most babies are born healthy regardless of a woman's age, obstetric history or family history.

• *Prenatal testing is an option available for you. Ensure you have sufficient information about your options to make an informed choice about testing.*

• *Most women receive reassuring test results that provide peace of mind in their pregnancy. However, some women experience anxiety with testing which you may consider when making a choice about testing.*

Approved by the Division of Midwifery, Calgary Health Region, April, 2007